

WES CRANIOFACIAL ANOMALIES DG 3.00

| <i>Gene</i> | <i>Median coverage</i> | <i>% covered >10x</i> | <i>% covered >20x</i> | <i>OMIM disease ID</i> |
|-------------|------------------------|--------------------------|--------------------------|------------------------|
| ACP4 | 97.1 | 97.2 | 88.8 | 617297 |
| ADAMTSL4 | 134.5 | 100.0 | 99.2 | 225100 |
| ALX1 | 169.2 | 99.7 | 97.1 | 613456 |
| ALX3 | 124.9 | 77.9 | 73.3 | 136760 |
| ALX4 | 137.7 | 100.0 | 99.3 | 168500 |
| AMBN | 192.8 | 99.8 | 98.5 | 616270 |
| AMELX | 96.4 | 99.9 | 96.8 | 301200 |
| AMER1 | 108.0 | 99.9 | 98.5 | 300373 |
| AMTN | 137.6 | 99.6 | 98.6 | 617607 |
| ANKRD11 | 118.7 | 96.1 | 93.5 | 148050 |
| ARHGAP29 | 168.5 | 99.5 | 98.0 | 8 |
| AXIN2 | 140.7 | 100.0 | 99.9 | 608615 |
| BCOR | 117.1 | 99.6 | 97.4 | 300166 |
| BMP2 | 178.0 | 100.0 | 100.0 | - |
| BMP4 | 178.6 | 100.0 | 100.0 | 607932 |
| CCBE1 | 81.2 | 99.8 | 98.8 | - |
| CDC45 | 162.6 | 99.8 | 98.5 | 617063 |
| CDON | 132.5 | 100.0 | 99.6 | 614226 |
| CDSN | 146.4 | 100.0 | 100.0 | 602593 |
| CHD7 | 158.7 | 100.0 | 99.5 | 214800 |
| COL11A1 | 113.4 | 96.2 | 92.8 | 154780;604841 |
| COL11A2 | 127.0 | 100.0 | 99.7 | 215150;277610 |
| COL2A1 | 125.9 | 100.0 | 99.7 | 108300 |
| COL9A1 | 159.8 | 100.0 | 99.2 | 614135 |
| COL9A2 | 103.5 | 99.9 | 99.0 | 614284;600204 |

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|---------|-------|-------|-------|---------------|
| COL9A3 | 109.1 | 98.7 | 95.5 | 120270 |
| COLEC11 | 178.1 | 100.0 | 100.0 | - |
| CTSK | 105.3 | 100.0 | 99.9 | 265800 |
| CYP26B1 | 148.9 | 100.0 | 99.9 | - |
| DHODH | 112.7 | 100.0 | 100.0 | 263750 |
| DISP1 | 190.7 | 100.0 | 99.9 | 2 |
| DLX3 | 135.0 | 99.9 | 98.4 | 104510 |
| DLX4 | 258.2 | 100.0 | 100.0 | 3 |
| DSPP | 67.8 | 96.8 | 86.1 | 125490 |
| EDA | 102.9 | 98.1 | 91.6 | 305100;313500 |
| EDAR | 148.4 | 100.0 | 99.9 | 129490;224900 |
| EDARADD | 101.9 | 99.9 | 98.8 | 129490;224900 |
| EDN1 | 166.4 | 100.0 | 100.0 | 615706 |
| EDNRA | 196.2 | 100.0 | 100.0 | 616367 |
| EFNA4 | 150.9 | 100.0 | 100.0 | 601380 |
| EFNB1 | 142.1 | 100.0 | 100.0 | 304110 |
| EFTUD2 | 123.9 | 100.0 | 99.8 | 610536 |
| EIF4A3 | 106.3 | 100.0 | 99.5 | 268305 |
| ENAM | 166.6 | 100.0 | 100.0 | 104500 |
| ERF | 144.8 | 99.9 | 98.5 | 600775 |
| ESCO2 | 139.5 | 98.7 | 95.2 | 268300 |
| EYA1 | 145.7 | 99.9 | 99.7 | 113650 |
| EZH2 | 171.4 | 100.0 | 99.5 | 277590 |
| FAM20A | 113.3 | 99.6 | 94.7 | 204690 |
| FAM83H | 76.6 | 84.9 | 81.5 | 130900 |
| FGD1 | 91.2 | 97.3 | 92.8 | 305400 |
| FGF10 | 155.3 | 100.0 | 99.8 | 149730 |
| FGF3 | 110.3 | 99.8 | 95.1 | 610706 |
| FGF8 | 127.7 | 98.2 | 88.9 | 4 |
| FGF9 | 225.5 | 100.0 | 100.0 | 612961 |

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|--------|-------|-------|-------|---------------|
| FGFR1 | 137.7 | 100.0 | 99.9 | 123150 |
| FGFR2 | 134.1 | 97.7 | 97.1 | 123500 |
| FGFR3 | 122.6 | 99.8 | 97.7 | 101400 |
| FLNA | 138.5 | 100.0 | 99.9 | - |
| FLNB | 140.5 | 99.5 | 98.8 | - |
| FOXC1 | 55.4 | 98.0 | 89.6 | 602482;601631 |
| FOXE1 | 42.5 | 96.9 | 78.5 | 241850 |
| GDF3 | 138.9 | 100.0 | 100.0 | 613702 |
| GDF6 | 116.8 | 100.0 | 99.9 | 118100 |
| GJA1 | 187.7 | 100.0 | 100.0 | 121014 |
| GJB6 | 145.5 | 100.0 | 100.0 | 129500 |
| GLI2 | 155.7 | 99.1 | 97.4 | 610829 |
| GLI3 | 140.8 | 98.5 | 98.0 | 175700 |
| GNAI3 | 108.8 | 99.3 | 95.2 | 602483 |
| GNPTAB | 167.2 | 100.0 | 99.9 | - |
| GPR68 | 138.4 | 99.5 | 96.7 | 617217 |
| GRHL3 | 156.5 | 100.0 | 100.0 | 606713 |
| GSC | 103.3 | 99.2 | 92.4 | 602471 |
| HOXA2 | 86.8 | 100.0 | 99.9 | 612290 |
| HUWE1 | 94.5 | 99.2 | 95.8 | 300706 |
| HYAL2 | 175.3 | 100.0 | 100.0 | 9 |
| IFT122 | 140.5 | 100.0 | 99.6 | 218330 |
| IFT43 | 134.3 | 100.0 | 100.0 | 614099 |
| IFT88 | 112.0 | 99.6 | 97.3 | 10 |
| IKBKG | 62.9 | 84.1 | 77.2 | 300291;300301 |
| IL11RA | 155.5 | 100.0 | 99.9 | 614188 |
| IL6ST | 112.4 | 96.4 | 90.3 | - |
| IMPAD1 | 157.7 | 100.0 | 100.0 | - |
| INTU | 141.9 | 99.7 | 98.1 | 617926 |
| IRF6 | 91.7 | 99.6 | 95.9 | 119300 |

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|---------|-------|-------|-------|----------------------|
| ITGB6 | 158.3 | 97.2 | 95.8 | 616221 |
| KAT6B | 171.3 | 99.6 | 98.3 | 606170;603736 |
| KDF1 | 111.2 | 100.0 | 99.8 | 617337 |
| KDM1A | 149.1 | 98.2 | 95.3 | 616728 |
| KDM6A | 114.8 | 96.1 | 88.7 | 300867 |
| KLK4 | 182.0 | 100.0 | 100.0 | 204700 |
| KMT2D | 142.1 | 100.0 | 99.4 | 147920 |
| KREMEN1 | 162.3 | 97.7 | 94.4 | 609898 |
| LAMB3 | 124.1 | 100.0 | 99.6 | 104530 |
| LRP2 | 171.0 | 100.0 | 99.9 | 222448 |
| LRP6 | 171.7 | 100.0 | 99.9 | 5 |
| LTBP3 | 132.5 | 99.6 | 98.1 | 613097 |
| MASP1 | 146.6 | 100.0 | 99.9 | 257920 |
| MED12 | 101.2 | 99.8 | 96.7 | 300895;305450;309520 |
| MEGF8 | 138.3 | 99.9 | 99.0 | 614976 |
| MEIS2 | 149.4 | 100.0 | 100.0 | 600987 |
| MEOX1 | 113.3 | 100.0 | 98.9 | 214300 |
| MID1 | 159.2 | 99.8 | 98.7 | 300000 |
| MITF | 160.6 | 100.0 | 99.9 | 193510 |
| MMP20 | 118.4 | 100.0 | 100.0 | 612529 |
| MN1 | 108.4 | 100.0 | 99.3 | - |
| MSX1 | 96.5 | 96.9 | 89.3 | 189500 |
| MSX2 | 90.4 | 100.0 | 99.4 | 168500 |
| NAA10 | 116.8 | 99.7 | 98.5 | 300013 |
| NECTIN1 | 135.3 | 100.0 | 99.9 | 225060 |
| NFKBIA | 132.6 | 95.2 | 88.0 | 612132 |
| NIPBL | 142.8 | 98.9 | 97.0 | 122470 |
| NOG | 175.6 | 100.0 | 100.0 | 186500 |
| NSD1 | 175.5 | 100.0 | 99.9 | 117550 |
| ODAPH | 279.7 | 100.0 | 100.0 | 614832 |

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|--------|-------|-------|-------|--------|
| OFD1 | 61.2 | 88.0 | 73.7 | 311200 |
| OTX2 | 152.3 | 100.0 | 99.7 | 610125 |
| P4HB | 119.0 | 94.6 | 94.0 | - |
| PAX3 | 114.5 | 100.0 | 99.9 | 193500 |
| PAX6 | 141.4 | 100.0 | 100.0 | 602482 |
| PAX7 | 135.1 | 100.0 | 100.0 | 268220 |
| PAX9 | 210.3 | 99.7 | 99.6 | 604625 |
| PGM1 | 147.7 | 94.2 | 94.2 | 614921 |
| PITX2 | 158.5 | 99.9 | 97.7 | 180500 |
| PLCB4 | 117.7 | 99.9 | 98.8 | 614669 |
| POLR1C | 102.8 | 90.5 | 87.0 | 248390 |
| POLR1D | 210.1 | 91.6 | 91.6 | 613717 |
| POR | 175.0 | 99.8 | 98.6 | - |
| PORCN | 121.0 | 100.0 | 99.1 | 305600 |
| PTCH1 | 127.2 | 99.2 | 97.6 | 109400 |
| PTH1R | 104.5 | 100.0 | 98.7 | 125350 |
| RAB23 | 130.8 | 100.0 | 99.5 | 201000 |
| RAD21 | 103.0 | 99.2 | 96.6 | 614701 |
| RBM10 | 121.7 | 99.5 | 97.1 | 311900 |
| RECQL4 | 149.8 | 99.8 | 98.1 | 603780 |
| RIPK4 | 144.6 | 100.0 | 99.9 | 263650 |
| RUNX2 | 113.5 | 72.2 | 72.2 | 119600 |
| SALL1 | 133.4 | 99.9 | 99.0 | 107480 |
| SALL4 | 136.9 | 98.6 | 96.7 | 607323 |
| SATB2 | 126.5 | 99.7 | 97.4 | 612313 |
| SCARF2 | 74.5 | 95.4 | 86.2 | - |
| SEC24D | 160.3 | 100.0 | 99.7 | - |
| SEMA3E | 158.1 | 99.2 | 98.9 | 214800 |
| SF3B4 | 69.4 | 99.9 | 97.3 | 154400 |
| SH3BP2 | 152.8 | 91.4 | 91.2 | 118400 |

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|---------|-------|-------|-------|---------------|
| SHH | 128.7 | 100.0 | 99.5 | 147250;611638 |
| SIX1 | 137.0 | 100.0 | 99.2 | 608389 |
| SIX3 | 154.3 | 99.9 | 98.6 | 157170 |
| SKI | 101.8 | 99.3 | 94.9 | 182212 |
| SLC24A4 | 123.9 | 100.0 | 99.8 | 615887 |
| SLC26A2 | 234.0 | 100.0 | 100.0 | 256050 |
| SMAD6 | 149.8 | 90.9 | 81.0 | 6 |
| SMC1A | 102.0 | 100.0 | 98.7 | 300590 |
| SMC3 | 91.6 | 95.2 | 91.0 | 610759 |
| SMO | 139.7 | 97.8 | 94.7 | 601707 |
| SMOC2 | 101.1 | 76.8 | 76.6 | 125400 |
| SNAI2 | 115.6 | 100.0 | 99.1 | 608890 |
| SOX10 | 70.3 | 99.9 | 97.9 | 613266 |
| SOX6 | 123.0 | 99.9 | 99.4 | 607257 |
| SOX9 | 161.0 | 100.0 | 98.6 | 114290 |
| SPECC1L | 135.8 | 96.0 | 95.7 | 600251 |
| SUMO1 | 23.7 | 67.2 | 49.9 | 613705 |
| TBX1 | 89.7 | 87.0 | 77.5 | 192430 |
| TBX22 | 142.6 | 99.2 | 95.7 | 303400 |
| TCF12 | 155.4 | 100.0 | 99.9 | 615314 |
| TCOF1 | 123.4 | 99.7 | 98.6 | 154500 |
| TFAP2A | 119.1 | 99.4 | 94.3 | 113620 |
| TGFBR1 | 181.8 | 93.7 | 93.6 | 609192 |
| TGFBR2 | 171.6 | 100.0 | 100.0 | 610168 |
| TGIF1 | 160.4 | 100.0 | 100.0 | 142946 |
| TLK2 | 108.4 | 99.1 | 95.1 | - |
| TP63 | 188.5 | 100.0 | 100.0 | 604292 |
| TRAF6 | 92.6 | 97.1 | 88.9 | 602355 |
| TSHZ1 | 155.0 | 98.8 | 98.8 | 607842 |
| TSPEAR | 160.3 | 100.0 | 99.2 | 618180 |

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|--------|-------|-------|-------|---------------|
| TWIST1 | 96.4 | 100.0 | 98.9 | 101400 |
| UBB | 49.1 | 100.0 | 99.4 | 119540 |
| VAX1 | 97.2 | 97.5 | 91.5 | 614402 |
| WDR19 | 153.1 | 100.0 | 99.4 | 614378 |
| WDR35 | 172.4 | 99.8 | 98.9 | 613610 |
| WDR72 | 151.9 | 96.8 | 96.4 | 613211 |
| WNT10A | 118.7 | 100.0 | 99.4 | 224750;257980 |
| WNT10B | 144.0 | 100.0 | 99.4 | 617073 |
| ZEB2 | 158.7 | 99.9 | 99.1 | 235730 |
| ZIC1 | 219.9 | 100.0 | 100.0 | 616602 |
| ZIC2 | 110.1 | 100.0 | 98.7 | 609637 |

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.

Median Coverage describes the average number of reads seen across 50 exomes.

% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors